# THPO gene

thrombopoietin

## Normal Function

The *THPO* gene provides instructions for making a protein called thrombopoietin that promotes the growth and division (proliferation) of cells. This protein attaches to (binds) and turns on (activates) the thrombopoietin receptor, which stimulates several signaling pathways that transmit chemical signals from outside the cell to the cell's nucleus. These pathways are important for controlling the production of blood cells.

Thrombopoietin is especially important for the proliferation of certain blood cells called megakaryocytes, which produce platelets, the cells involved in blood clotting. Research suggests that thrombopoietin signaling may also play a role in the renewal of hematopoietic stem cells, which are stem cells located within the bone marrow that have the potential to develop into red blood cells, white blood cells, and platelets.

## Health Conditions Related to Genetic Changes

### essential thrombocythemia

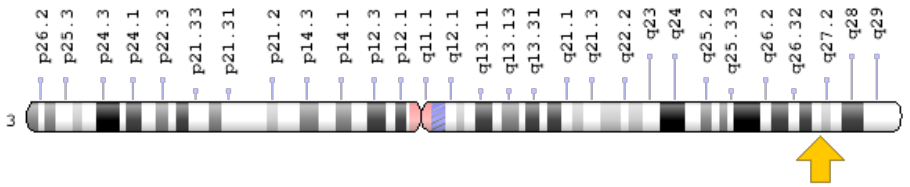
Several mutations in the *THPO* gene have been found in people with essential thrombocythemia, a condition characterized by an increased number of platelets in the blood. Because platelets are the blood cells involved in blood clotting, abnormal clotting (thrombosis) is common in people with essential thrombocythemia.

*THPO* gene mutations are found in families with an inherited form of the condition called familial essential thrombocythemia. These mutations affect a region of the gene that usually blocks (inhibits) the production of the thrombopoietin protein (a process called translation). *THPO* gene mutations lead to increased translation of the protein. The excess protein can abnormally activate the thrombopoietin receptor and the signaling pathways, leading to overproduction of megakaryocytes and increased numbers of platelets. Excess platelets can cause thrombosis, which leads to many signs and symptoms of essential thrombocythemia.

## Chromosomal Location

Cytogenetic Location: 3q27.1, which is the long (q) arm of chromosome 3 at position 27.1

Molecular Location: base pairs 184,371,935 to 184,379,688 on chromosome 3 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- c-mpl ligand
- megakaryocyte colony-stimulating factor
- megakaryocyte growth and development factor
- megakaryocyte stimulating factor
- MGC163194
- MGDF
- MKCSF
- ML
- MPL ligand
- MPLLG
- myeloproliferative leukemia virus oncogene ligand
- TPO
- TPO_HUMAN

## Additional Information & Resources

Educational Resources

- Holland-Frei Cancer Medicine (6th Edition, 2003): Thrombopoietin https://www.ncbi.nlm.nih.gov/books/NBK12518/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28THPO%5BTIAB%5D%29+OR+%28thrombopoietin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- THROMBOPOIETIN
  http://omim.org/entry/600044

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_THPO.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=THPO%5Bgene%5D
- HGNC Gene Family: Endogenous ligands
  http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11795
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/7066
- UniProt
  http://www.uniprot.org/uniprot/P40225

## Sources for This Summary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